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Abstract

Coronary Heart disease (CHD) is the biggest killer in the United Kingdom. In 2018, over one quarter of deaths were due to CHD (British Heart Foundation, 2018), with congenital defects identified as the most common cause of heart disease in pregnancy (Wylie & Bryce, 2016).

This article will focus on Marfan syndrome and the impact this has on pregnancy and childbirth. Current literature and research will be appraised and discussed to explore mode of delivery during the second stage of labour and calculate the most appropriate method of delivery. Additionally, this article will address how the midwife can further support women with Marfan syndrome during the pregnancy booking, antenatal period and intrapartum period without labelling them, and discuss how this may be achieved in relation to the uncertainty reduction communication theory.

Marfan Syndrome

Marfan syndrome (MFS) was first described by the French doctor Bernard J.A. Marfan in 1896, and is a hereditary autosomal dominant disorder of connective tissue affecting collagen and elastin within a variety of body systems such as musculoskeletal, cardiovascular, respiratory, ocular and integumentary systems (Keane and Pyeritz, 2008).

The Marfan Foundation (2018) states that the incidence of MFS in the UK population is approximately 18,000, with 200 new cases diagnosed every year in the UK and worldwide, statistics show that 1 in 3,300 are affected by MFS with approximately 50% of sufferers remaining undiagnosed.

The disease process by which MFS presents itself, is identified by a mutation in the gene for fibrillin-1 on chromosome 15. There are more than >1000 mutations and each one is unique

to an individual or family (Keane and Pyeritz, 2008). If one parent has MFS, there is a 50% risk of the fetus inheriting the mutant gene (Robson and Waugh, 2013). Indeed, Gambling *et al.* (2008) further support this by examining the incidence of MFS, suggesting 25% of diagnosed cases arise as new mutation. Consequently, this indicates further research would be beneficial, as the correlation between the genotype- phenotype of MFS is unclear due to the large number of unique mutations.

There are a multitude of signs and symptoms of MFS; the visual appearance of a patient with the condition can present as tall, with a thin physique, very little subcutaneous fat and with an excessive length of long bones, long fingers and toes (Robson & Waugh, 2013). Also, Swan (2006) suggest that while both men and women are affected by the condition equally, it may appear more difficult to diagnose women as the appearance is less exaggerated than with their male counterparts. This could lead to a plethora of complications if the woman was to enter pregnancy undiagnosed, as many symptoms of cardiac disease are similar to those of pregnancy, this will be discussed later in this paper.

Associated general complications of MFS are outlined by Kumar & Clarke (2016) as being increased joint pain and possible dislocations due to the laxity of joints. Additionally, those with MFS may encounter hernias, fatigue, and scoliosis of the spine leading to restrictions with mobility. Also, spontaneous pneumothorax may be experienced in more than 10% of those affected with MFS. However, one of the most severe complications of the disease itself are cardiac complications. The main concern of the cardiovascular system for those with MFS is the progressive enlargement of the aortic root diameter. The measurement of the aortic root diameter is of great importance during pregnancy as Brooke *et al* (2008) suggests, as progressive enlargement of the aortic root can lead to dissection which accounts for 20% of maternal cardiac fatalities. A normal aorta root diameter of the heart

measures between 20mm and 35mm but, as there is progressive enlargement of the aortic root with those affected with MFS, it is essential there is an early diagnosis of the condition so there is medical management within a detailed care plan. Furthermore, it is crucial women with MFS receive regular echocardiography, a multi-disciplinary assessment and discussions regarding lifestyle choices such as healthy eating, exercise, and smoking cessation and these are addressed during pre-conception care, so the woman has achieved optimal health before embarking on pregnancy (Robson & Waugh, 2013). Also, Pre-conception counselling, genetic counselling and advice regarding treatments should be considered, so women are aware of the risks posed to her and her baby during the pregnancy, specifically as use of beta-blockers are advised throughout pregnancy to reduce aortic root dilatation (Marfan Foundation, 2018).

Pregnancy Booking

The role of the midwife in relation to cardiac disease in pregnancy is of utmost importance. The midwife must be aware of signs and symptoms related to cardiac problems throughout the pregnancy, and make referrals to other professionals within a multi-disciplinary team when necessary and as quickly and as effectively as possible without delay.

Women who present at booking with a known cardiac problem must be considered high risk and therefore have shared care which includes the midwife, obstetrician, cardiologist and anaesthetist (Wylie & Bryce, 2016). For women who present later in pregnancy with a suspected cardiac problem, diagnosis of the condition is usually based on tests such as blood tests, an electrocardiogram, echocardiography and chest X-rays all of which are considered safe in later pregnancy (Lewis & Drife, 2004). Thus, the role of the midwife is to

take a comprehensive booking history and promptly make referrals to other health professionals. Respiration and pulse must be monitored, as well as other vital observations at each antenatal visit, and women encouraged to take iron and vitamins to prevent anaemia which can induce additional stress on the heart due to lack of oxygen in the body (Burrow *et al*, 2004).

Wylie & Bryce (2016) indicate the difficulty of recognising symptoms of cardiac problems in pregnancy, as pregnancy can produce 'similar responses' in the body'. Therefore, the midwife must have full knowledge of the woman's baseline observations so possible complications can be identified which are unrelated to pregnancy. A New York Classification system (Table 1) was developed to identify the extent of heart failure which works by assessing the woman and placing her in one of four categories. These categories are based on the woman's limitations during physical activity and her symptoms, with regards to normal breathing, shortness of breath, angina pain and palpitations (Gambling *et al*. 2008). It could be considered the New York Classification system is a useful tool which could be used by midwives, to assist in identifying the degree of strain on the heart due to the additional workload during pregnancy.

The impact of pregnancy has a significant risk on the compromised heart, and as suggested by O'Brien & Walker (2014) there are three periods during the childbearing process which are considered as critical. Between 28-32 weeks gestation, there are haemodynamic changes which reach their peak during this stage in the pregnancy due to a 50% increase in blood volume, combined with the effects of pregnancy hormones which cause connective tissue changes and an increase of blood vessels throughout the uterus, all of which increase the workload of the heart. Also, during labour there is an increase in cardiac output due to physical stress of uterine contractions and psychological stress of labour pain; and finally 12-

24 hours postpartum is considered a critical period as additional blood volume from the uterus and placental site return to the systemic circulation. Consequently, due to the increased cardiac output during these three critical periods, there is an increased risk of aortic dissection during pregnancy due to additional stress placed on the aortic root, and as suggested by Fujitani & Baldisseri (2005) 50% of aortic dissections in women occur during pregnancy.

Although most women who present with cardiac disease can have a positive outcome of pregnancy according to Davis & Macnab (2016), in contrast Gambling *et al.* (2008) suggest those women who have an aortic root of more than 40mm should be advised against pregnancy irrespective of their New York Classification as they are at greater risk of aortic dissection. An aortic root of more than 40mm carries a fatality rate of 36-72% within 48 hours, and without intervention such as surgery or a chronic beta-blocker therapy, death will become imminent within one week. Therefore, pre-conception advice and full physical examination are crucial for those with cardiac disease, alongside a detailed care plan which is regularly evaluated and updated within a multi-disciplinary team to ensure a positive outcome of pregnancy (Robson & Waugh, 2013).

Furthermore, additional antenatal appointments should be arranged in consultation with the Obstetrician, ideally where all professionals are available in one clinic which would reduce the amount of visits required and advice given regarding compliance of prescribed drugs to avoid possible further complication (Kumar & Clark, 2016). A pre-delivery anaesthetic assessment is recommended, as Gambling *et al.* (2008) suggest women with MFS can experience potential airway management problems due to cervical spine instability and a high arched palate. Kumar & Clark (2016) also note, women should have regular echocardiograms at six week intervals to identify any changes to the aortic root diameter,

whilst according to a study by Rosenblum *et al.* (1983) even regular echocardiograms may fail to 'predict' aortic dissection in pregnancy for those women who have MFS. However, the reliability of the Rosenblum *et al.* (1983) study could now be questioned as it is outdated and advances in technology now mean echocardiograms could be considered a good predictor of possible dissection (Burrow *et al.* 2004). During the antenatal period, fetal growth will be regularly assessed by ultrasound, and an anomaly scan will be advised at 16-18 weeks gestation to detect for congenital abnormalities, as the risk of a cardiac defect is increased in these babies (Nanda *et al.*, 2012). Furthermore, it is suggested the midwife should advise the woman of possible premature labour and ruptured membranes as this is common with those who have MFS (Meijboom *et al.* 2006).

Mode of Delivery and Intrapartum Care

It is vitally important, a plan of care is made well in advance regarding mode of delivery, timing and type of analgesia and anaesthesia, and these decisions should be made within the multi-disciplinary team in discussion with the woman (Yu-Ling Tan, 2010). During labour and delivery, there are significant haemodynamic changes which occur with each uterine contraction which can be tolerated by healthy women however, for those with heart disease it can result in morbidity and mortality. This is due to a 300-500ml transfer of blood back into the systemic circulation with each contraction, in addition, alongside the anxiety of labour and the sympathetic response to pain, there is an increase in both heart rate and blood pressure therefore increasing cardiac output by 34% during contractions and 12% between contractions; this can lead to greater stress on the enlarged aortic root, ultimately increasing the risk of aortic dissection (Meijboom *et al.* 2006). For this reason, it is important to examine a range of research and literature exploring vaginal delivery and caesarean section, to calculate the most appropriate method of delivery.

It has been suggested by Langesaeter *et al.* (2009), there is a greater focus on caesarean section as preferred mode of delivery rather than vaginal delivery. Caesarean section can be advantageous in respect of date of delivery, and anaesthesia can be planned well in advance so further risk of damage or dissection to the aortic root can be avoided, however it could be questioned for who is it advantageous. In a Dutch study by Meijboom *et al.* (2006) which investigated the obstetric maternal outcome of pregnancy in women with Marfan syndrome, it suggested out of the 122 enrolled women 31% had a caesarean section compared to the Dutch general population of 11% which questions whether the decision for caesarean section was in the best interests of the woman, or if the high rate of caesarean reflects fear and caution for further complications by obstetricians and cardiologists. Furthermore, the study did not take into consideration the woman's New York classification of cardiovascular disease which further questions the reasons for opting for caesarean as it is suggested by Wylie & Bryce (2016) women who are considered to be class one or class two of the New York classification can often experience quick, uncomplicated labours and as long as the woman is given adequate analgesia to keep physiological and psychological stress minimised, a vaginal delivery can be achieved. Moreover, Meijboom *et al.* (2006) suggest there are greater risks with caesarean such as fluctuations of blood pressure as a result of intubation and anaesthetic agent for those who require general anaesthetic; increased risk of wound and uterine infection, also greater risk of venous thromboembolism and increased blood loss compared with vaginal delivery. However, Langesaeter *et al.* (2009) defends caesarean and suggests it is the safest mode of delivery as haemodynamic stability can be achieved through use of regional anaesthesia such as epidural, as cardiac output is decreased therefore causing vasodilation which reduces venous resistance, alongside use of intravenous doses of oxytocin and invasive monitoring such as an arterial line.

According to Richards & Yentis (2006), vaginal delivery is the safest mode of delivery as it causes less stress to the heart however, there are a number of considerations to be taken into account when a vaginal delivery is planned. During the first stage of labour, it is advisable for the woman to remain in an upright or left lateral position to prevent aortocaval compression therefore displacing the uterus away from the aorta and vena cava. However most importantly, directed pushing during the second stage of labour must not be encouraged as this causes a decrease and sometimes a complete loss in cardiac output which cannot be restored (Richards & Yentis, 2006). Robson & Waugh (2013) could be criticised for suggesting directed pushing during the second stage of labour, as they fail to question the consequences of such an action in relation to the stress it would place upon the heart, therefore leading to possible aortic dissection which consequently accounts for 20% of maternal cardiac fatalities (Swan, 2006). Furthermore, Robson & Waugh (2013) fail to provide evidence to support their recommendation of directed pushing in relation to women with cardiac disease in pregnancy. Also, it is important to note, some women may require an assisted vaginal delivery whereby Meijboom *et al.* (2006) suggests vacuum or forceps be used, so long as effective analgesia was utilised such as epidural early in labour, as this would minimise physiological stress and outweigh any risk compared with those of caesarean section.

Following analysis of a range of material in support of both vaginal delivery and caesarean section, it could be argued the most appropriate mode of delivery is vaginal delivery with spontaneous pushing and if required, with aid of instrumental delivery as the risks associated with this outweigh any risk associated with caesarean section. However, it is important to highlight women with cardiac disease in pregnancy, must always give birth in an obstetric unit where all professionals within the multi-disciplinary team are available. Also, close monitoring of vital signs is crucial, alongside use of an echocardiography throughout labour and someone who can interpret the findings, likewise, oxygen therapy should be

available if required, as well as close monitoring of fluid balance through the use of a central venous pressure catheter if necessary (Roberts & Ketchell, 2012). Furthermore, it was found when analysing mode of delivery, there does not seem to be a definite appropriate method as most of the research and articles contradicted one another, which indicates the decision of mode of delivery needs to be made in relation to individual circumstances in discussion with the woman.

How Can Midwives Support Women?

Caring for women with cardiac disease in pregnancy can be considered outside the parameters of the midwife and a need for referral to other practitioners is essential. Moreover, as expressed by the NMC (2018) the midwife will continue to give care as part of a team and will need to be aware of any cardiac compromise throughout pregnancy and childbirth. Therefore, it could be questioned how midwives can best support women with cardiac disease.

Within midwifery practice, women-centred care remains paramount and so the midwife should encourage a two-way transfer of knowledge and information at each contact. This would ensure a trusting relationship with the woman, so midwives are able to organise a plan of care based on the information she herself has knowledge of. Also, the importance of gradually 'drip-feeding' new information to the woman is essential, so she is able to acquire her own knowledge which will empower her to make decisions about her own care, and allow her to prepare for critical periods during pregnancy and childbirth.

According to West & Turner (2018) uncertainty can cause stress and anxiety, so by examining communication theories, in particular the uncertainty reduction theory in seminal literature by Berger (1979), it can be understood that good communication is the foundation of how midwives can support women with heart disease. It is thought, "One of the motivations underpinning interpersonal communication is the acquisition of information with which to reduce uncertainty" (Heath and Bryant, 2013, p.259). Thus, it is vital midwives discuss all options concerning care with women and include them in decision making; they act as an advocate for them, and offer information so they can make an informed decision, to prepare them for the possible need of hospital care in the third trimester of pregnancy. It can be considered difficult when trying to normalise pregnancy for women with heart disease without labelling them. Therefore, it is important the woman is invited to parent craft classes alongside other pregnant women so she feels included, whereby health promotion can be discussed with everyone such as the advantages of a well-balanced diet and controlling weight gain, regardless of the fact she may have already discussed this with another professional involved in her care.

Conclusion

In conclusion, a multi-disciplinary approach is essential for successful management of those who present in pregnancy with cardiovascular disease. Also, the midwife must be aware of the signs and symptoms of CHD for those women who are unaware they have a cardiac problem and make immediate referrals to other professionals. The New York Classification of cardiovascular disease is a useful tool which can be used by midwives, to inform practice by alerting them to any additional stress which is placed on the heart. Women-centred care is essential within midwifery practice and by sharing knowledge and information of MFS with the woman, midwives and other health professionals are able to organise a plan of care

through joint decision-making, which in turn will empower her and prepare her for critical periods during pregnancy and childbirth. In general, vaginal delivery with epidural analgesia is the preferred mode of delivery compared with caesarean section as there are fewer risks and haemodynamic stability can be achieved. Moreover, by examining communication theories such as the uncertainty reduction theory, it was found good communication is the foundation of the support midwives can give to women who have cardiovascular problems in pregnancy and by inviting these women to sessions such as parent craft to discuss health promotion as a larger group, it may help women feel included, therefore attempting to normalise her pregnancy.

New York Classification of Cardiovascular Disease.

Classification	Patient Characteristics
Class One: No objective evidence of cardiovascular disease.	Patients with cardiac disease but without resulting limitation of physical activity. Ordinary physical activity in the patient does not cause fatigue, palpitation, dyspnoea or angina.
Class Two: Objective evidence of minimal cardiovascular disease.	Patients with cardiac disease resulting in slight limitation of physical activity. The patient is comfortable at rest. Ordinary physical activity results in fatigue, palpitation, dyspnoea or angina.
Class Three: Objective evidence of moderately severe cardiovascular disease.	Patients with cardiac disease resulting in marked limitation of physical activity. They are comfortable at rest. Less than ordinary activity causes fatigue, palpitation, dyspnoea or angina.
Class Four: Objective evidence of severe cardiovascular disease.	Patients with cardiac disease resulting in an inability to carry on any physical without discomfort. Symptoms of heart failure or angina may be present even when the patient is at rest. If physical activity is undertaken, discomfort increases.

Adapted from Wylie & Bryce (2016) *The Midwives Guide to Key Medical Conditions*.

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Keywords:

Marfan Syndrome; Chronic Heart Disease; Chronic Heart Disease in Pregnancy; Mode of Delivery for Women with Marfan Syndrome; Uncertainty Reduction Communication Theory; Midwives Support for Women with Heart Disease.

Key points:

- Marfan syndrome in the UK incidence of approximately 18000, with 200 new cases diagnosed every year in the UK.
- A plethora of complications can occur if a woman was to enter pregnancy with heart disease undiagnosed, as many symptoms are similar to those of pregnancy.
- It is suggested, the most appropriate mode of delivery is vaginal delivery, as the risks associated with this outweigh any risk associated with caesarean section.
- Women with coronary heart disease in pregnancy may feel excluded due to medicalisation of her care. Midwives can support women and help inclusion by inviting them to parent craft sessions to discuss health promotion as a larger group, therefore attempting to 'normalise' her pregnancy.

Reflective Questions:

- How would you ensure women with CHD feel empowered during their pregnancy? How would you advocate for her whilst in your care?
- How would you support women with CHD who could potentially decline/ not attend regular medical appointments with multidisciplinary colleagues during pregnancy?
- What is your referral pathway, if you suspect a woman in your care presents with cardiac problems? Could you recognise these alongside normal pregnancy symptoms? How would you express your concerns to the woman without distressing her?